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Leukaemia Section
Short Communication

t(6;21)(p22;q22)

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Clinics and pathology

Disease

Treatment related myelodysplastic syndrome (refractory anemia with excess of blasts: RAEB).

Etiology

RAEB occurred 60 w after diagnosis of an acute lymphoblastic leukemia treated with topoisomerase II inhibitors.

Epidemiology

Only one case to date, a 4 yr old female patient.

Prognosis

The patient died 10 mths after diagnosis.

Cytogenetics

Cytogenetics morphological

a t(2;11)(p23;q23) with MLL involvement was also present in the same clone.

Genes involved and proteins

Note

The gene in 6p22 is yet unknown, and, because cryptic t(12;21) ETV6 /AML1 are not rare, it is therefore uncertain whether this translocation involve a new AML1 partner.

AML1

Location

21q22

DNA/RNA

Transcription is from telomere to centromere

Protein

Contains a Runt domain and, in the C-term, a transactivation domain; forms heterodimers; widely expressed; nuclear localisation; transcription factor (activator) for various hematopoietic-specific genes

References

Mathew S, Head D, Rubnitz JE, Raimondi SC. Concurrent translocations of MLL and CBFA2 (AML1) genes with new partner breakpoints in a child with secondary myelodysplastic syndrome after treatment of acute lymphoblastic leukemia. Genes Chromosomes Cancer. 2000 Jun;28(2):227-32

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